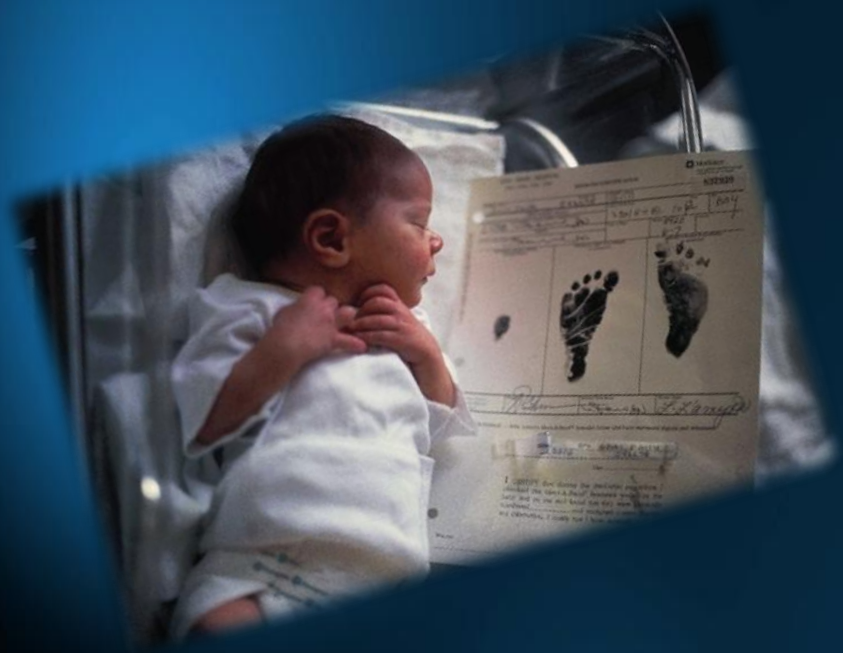
NEWBORN SCREENING IN NEBRASKA

Newborn Screening for Metabolic & Inherited Disorders

and

Early Hearing Detection & Intervention



2010 Annual Report

Department of Health & Human Services

DHHS

NEBRASKA

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NEBRASKA EARLY HEARING DETECTION AND INTERVENTION ANNUAL REPORT - 2010

NEWBORN SCREENING FOR INBORN ERRORS OF METABOLISM AND INHERITED DISORDERS

The goal of newborn blood spot screening is to identify newborns at risk for certain metabolic, endocrine, hematologic and other conditions that would otherwise be undetected until damage has occurred, and for which intervention and/or treatment can improve the outcome for the newborn.

Newborn Screening is a system involving many elements including:

- Education of health care professionals and parents and efforts to increase public awareness
- Proper and timely collection of quality specimens
- Appropriate and timely transmittal of specimens to the Newborn Screening laboratory
- Rapid quality testing methods
- Timely notification of the infant's physician and parents
- Timely recall of the infant for confirmatory or repeat testing
- Appropriate referral of family to specialists for diagnosis, treatment and counseling
- Assuring access to needed specialized services and treatment
- Evaluation and Quality Assurance

Each of these components of the system requires ongoing monitoring to ensure quality.

In 2010, newborn screening efforts resulted in successfully identifying and treating 40 newborns affected with conditions in time to prevent problems associated with them:

- 1 baby with argininosuccinic acidemia (ASA)
- 1 baby with partial (treated) biotinidase deficiency (BIO)
- 2 babies with congenital adrenal hyperplasia (CAH)
- 11 babies with congenital primary hypothyroidism (CPH)
- 4 babies with cystic fibrosis
- 1 baby with Duarte Galactosemia
- 8 babies with hemoglobinopathies (5 sickle cell disease, 1 SC-disease, 2 C-disease, and 1 hemoglobin C beta thalassemia)
- 1 baby with hypermethioninemia
- 1 baby with long chain hydroxyl acyl-CoA dehydrogenase deficiency (LCHAD)
- 1 baby with medium chain acyl-coA dehydrogenase deficiency (MCAD)
- 3 babies with methylmalonic acidemia (MMA)
- 2 babies with phenyketonuria (PKU) + 2 benign hyperphenylalaninemia
- 1 baby with short chain acyl-CoA dehydrogenase deficiency (SCAD)
- 1 with LCHAD (long chain hydroxy acyl-coA dehydrogenase deficiency (LCHAD)
- 7 babies with transient tyrosinemia (not included in count above)

The incidence rate of conditions in Nebraska based on the screened conditions identified from 2006 - 2010 and number of births screened those five years: 1:604 births

WHAT IS NEWBORN SCREENING?

Newborn screening programs have been around for over four decades in all 50 states and in several countries. The compulsory screening panel varies slightly from state to state but the overall goal is the same: prevent or minimize the serious effects of the conditions screened. In 2010 Nebraska's required screening panel included 28 metabolic, endocrine, hematologic and other conditions.

The effects of screened conditions if not detected and treated can range from brain and nerve cell damage resulting in severe intellectual disability, to damage to the child's heart, kidney, liver, spleen, eyes, problems with physical growth, stroke and even death.

The conditions for which screening is done, are individually rare, so consultation with and/or referral to the appropriate pediatric specialist such as a geneticist, metabolic specialist, hematologist, endocrinologist or an Accredited CF Center is always recommended.

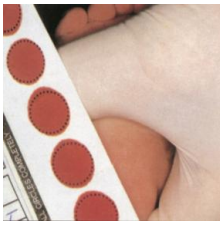
Conditions included in Nebraska's required blood-spot screening panel in 2010 were:

- Arginino Succinic Acidemia
- Beta-ketothiolase Deficiency
- Biotinidase Deficiency
- Carnitine Uptake Defect
- Citrullinemia
- Congenital Adrenal Hyperplasia
- Congenital Primary Hypothyroidism
- Cystic Fibrosis
- Galactosemia
- Glutaric Acidemia Type I
- Hemoglobinopathies
  (Sickle Cell, Hgb. C & Thalassemias)
- Homocystinuria
- Isovaleric Acidemia
- Maple Syrup Urine Disease
- Long Chain Hydroxy Acyl-CoA Dehydrogenase Def.
- Medium Chain Acyl-CoA Dehydrogenase Deficiency
- Methylmalonic Acidemia (Mutase)
- Methylmalonic Acidemia (Cbl A & B)
- Multiple Carboxylase Deficiency
- Phenylketonuria
- Propionic Acidemia
- Tyrosinemia
- Trifunctional Protein Deficiency
- Very Long Chain Acyl-CoA Dehydrogenase Deficiency
- 3-Hydroxy 3-Methyl Glutaric Aciduria
- 3-Methylcrotonyl-CoA Carboxylase Deficiency

HOW THE NEWBORN SCREENING PROCESS WORKS

1: TESTING

Baby is born.
Dried blood spot specimen is collected @ 24-48 hours of life



Specimen shipped overnight to newborn screening laboratory, PerkinElmer

Specimen data entered into data system

Specimen tested for multiple conditions

2: FOLLOW UP

Inconclusive or positive screen results reported by phone/fax/letter from lab and State Program staff.

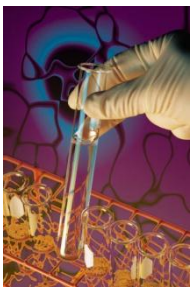
Baby's physician or health care provider contacts baby's parents

Parent's bring baby back in for evaluation and more testing

3: DIAGNOSIS/ INTERVENTION

If screening results indicate a need:

Repeat or confirmatory testing occurs



Parent education on signs/symptoms to watch for

Baby's physician consults with and/or refers baby to pediatric sub-specialist appropriate to the condition

4: TREATMENT & MANAGEMENT

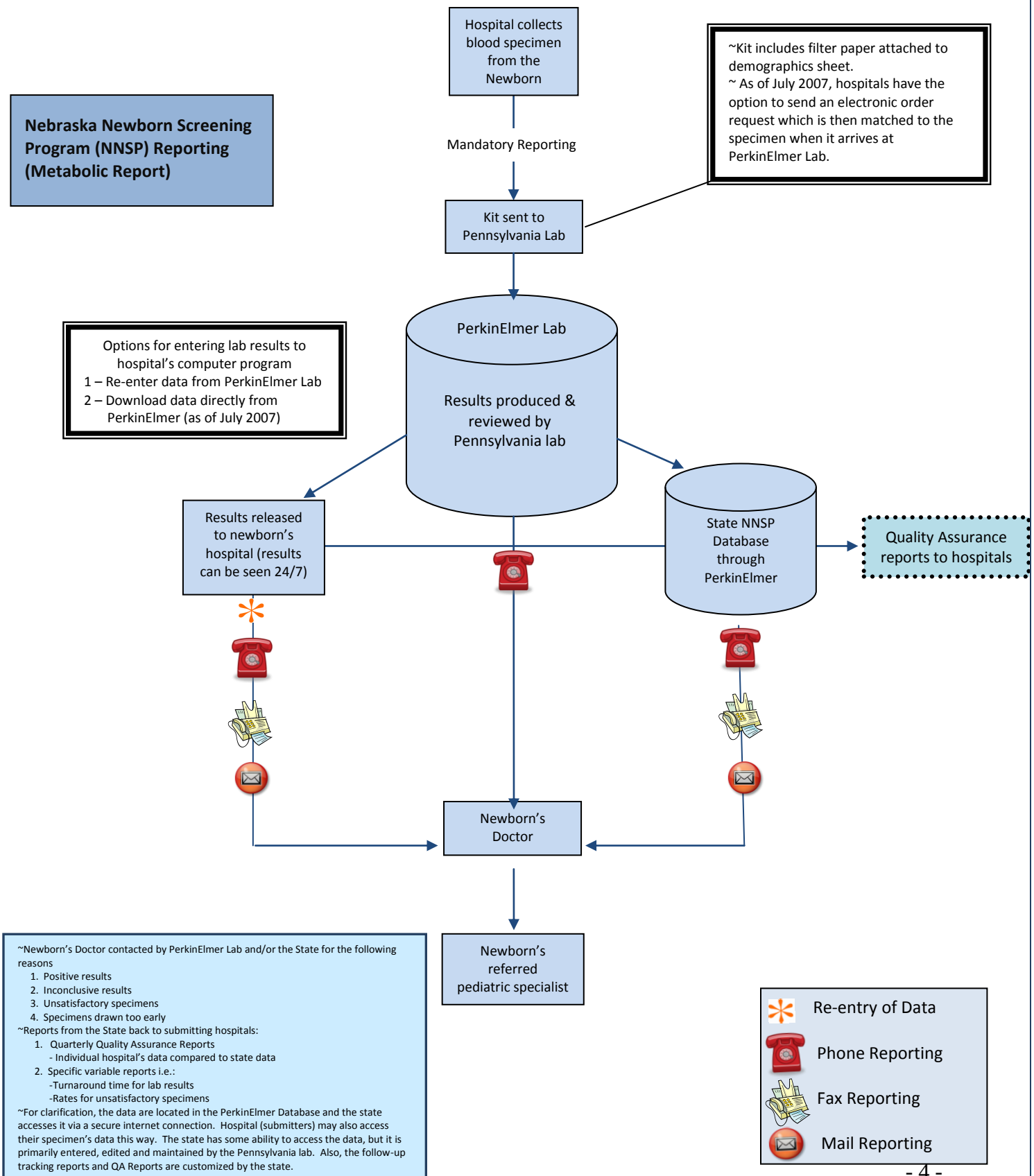
Once diagnosis is made, treatment begins. (For some life threatening conditions, treatment may occur prior to diagnosis- on recommendation of specialist.)

Parents receive treatment instructions / education.

Team Support services as appropriate, e.g.:

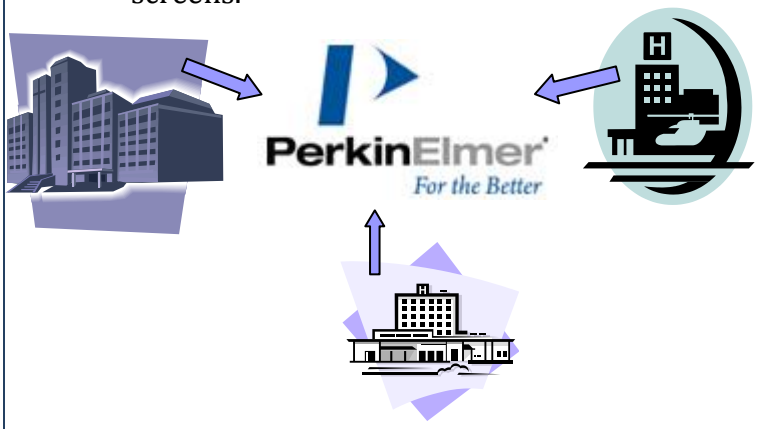
- metabolic dietitian monitoring & consultation
- ongoing blood monitoring
- referral to early intervention services
- pulmonary/ CF services
- ped endocrine monitoring
- ped hematology monitoring
- genetic counseling & consideration of family testing
- Other allied health services as needed

Data Flow: **This chart demonstrates how data from newborn screening is produced, transmitted and utilized to facilitate the recall of newborns at risk for any of the screening panel conditions. They can then be evaluated, diagnosed and have treatment initiated.**



System Overview

In 2010, 59 Nebraska hospitals sent specimens to PerkinElmer Screening Laboratory. This laboratory is under contract with the State of Nebraska to conduct all of the newborn screens.



The Newborn Screening Program in the Nebraska Department of Health and Human Services was staffed by Mike Rooney, Administrative Assistant, Krystal Baumert, Follow-up Coordinator, Karen Eveans, Follow-up Specialist, and Julie Luedtke, Program Manager.

Expert advice and assistance were available as needed throughout the year by consultation with the laboratory staff and other specialists. The specialists in metabolic diseases were Richard Lutz, M.D., William Rizzo M.D., Jill Skrabal, R.D., Kathryn Heldt, R.D., and Rose Kreikemeier, RN. Consultation regarding Cystic Fibrosis were with the CF Center Director John Colombo, M.D. and Dee Aquazzino CF Center Coordinator. Pediatric endocrinologist Kevin Corley, MD, and pediatric hematologist James Harper, M.D. were frequently consulted.

Quarterly meetings with the Newborn Screening Advisory Committee provided invaluable guidance to the program on several policy and quality assurance issues.

Treatment services received support via the $10 per infant screened fee, State General Funds and Title V Maternal and Child Health Block Grant funds. This included funding for special metabolic formulas, metabolically altered/pharmaceutically manufactured foods, and support for specialty dietitian services and sub-specialist M.D. consultation services.

Quarterly quality assurance reports were sent to every birthing hospital, as well as Children's Hospital of Omaha, a facility that completes a significant number of screens on babies transferred to them. In addition, the Advisory Committee reviewed several quality assurance reports at each quarterly meeting.

MAJOR INITIATIVES of 2010 in NEBRASKA

Education

- ❖ Mike Rooney of the Nebraska Newborn Screening Program continued to track and distribute the "Parents Guide To Your Baby's Newborn Screening" to the 59 birthing hospitals, Children's hospital and upon request to some Obstetric, Family Physician and Pediatric practices.

- ❖ Local presentations by the program manager included a newborn screening update for the Physician Seminar Day at St. Elizabeth Regional Medical Center in Lincoln, the ASCP/CLMA/NSCLS (laboratory) Spring meeting and the Central Plains Society of AMT's Fall meeting. She also presented at the national NBS & Genetics Symposium with Judi Tuerck, RN, MSN on the guidelines for Newborn Screening of Premature Low Birth Weight and Sick Newborns, as well as a national teleconference sponsored by CLSI on these guidelines.

- ❖ Internal staff development efforts included the Program Manager and follow-up staff attending the Association of Public Health Laboratory's National NBS & Genetics Symposium in Orlando, and the Heartland NBS & Genetics meeting in Des Moines. Karen Eveans attended a national meeting on hemoglobinopathy screening.

- ❖ Educational mailings to prepare pediatricians, family physicians, neonatologists, hospital laboratory and nursery personnel were sent out in December. This addressed the recommendations for serial screening of NICU admissions of premature, low birth weight and sick newborns.

Policy

- Newborn Screening Program staff Krystal Baumert and Karen Eveans continued to serve on the Newborn Screening Committee of the Heartland Newborn Screening and Genetics Collaborative. Karen Eveans served as an advisor to the CLSI workgroup developing guidelines for screening for cystic fibrosis.

- The program manager continued to serve on: the APHL's Newborn Screening & Genetics Committee, the Heartland Region's Advisory Council, and the Newborn Screening workgroup, and the National Coordinating Centers Long Term Follow-up Work Group.

- The Newborn Screening Advisory Committee continued its quarterly review of quality assurance data of preanalytical (e.g. unsatisfactory specimen rates and types), analytical (e.g. statistical performance of assays over time) and post-analytical (e.g. age at time of intervention or treatment for diagnosed patients) performance measures for the system.

- The Newborn Screening Advisory Committee reviewed and evaluated several technical issues and changes proposed by the newborn screening laboratory relative to screening algorithms for cystic fibrosis and congenital adrenal hyperplasia. The Committee also

approved new information to be placed on newborn screening test results for babies who had abnormal screen results and were low birth weight, to provide physicians' additional information based on birth weight ranges.

- The Committee continued its evaluation of Severe Combined Immune Deficiency as a candidate condition for screening. The Secretary of Health and Human Services endorsed the recommendation by the Secretary's Advisory Committee on Heritable Diseases in Newborns and Children to make SCID part of the Recommended Universal Screening Panel (RUSP) in 2010.

- In response to the NCAA policy on identifying sickle cell trait status in collegiate athletes, inquiries to the screening program for test results were increasing. In response to this, the policy on release of information was clarified and approved by the NBS Advisory Committee.

- In response to the publicity surrounding lawsuits in Minnesota and Texas regarding the use of left-over dried blood spots from newborn screening, the program clarified its policy and the regulations on how those may be used. This information was posted on Nebraska's home page of its NBS web-site.

- The Newborn Screening Advisory Committee (NBSAC) undertook evaluation of proposed regulations recommending revisions to sections addressing:
    - the storage, use and disposal of residual newborn dried blood spots,
    - the quality of information to be included on confirmatory laboratory test results,
    - adoption of the current edition of the Clinical and Laboratory Standards Institute standards for blood collection which would allow with certain precautions the collection of blood spots via umbilical catheter for newborns in the neonatal intensive care unit, and
    - clean-up language and clarifying definitions.

These regulation revisions underwent the public regulatory review process in 2010.

- The NBSAC in collaboration with the Advisory Committee for the EHDI program continued its evaluation of the multiple policy implications of various models of integrating dried blood spot testing for Congenital Cytomegalovirus (CMV) and other genetic causes of hearing loss.

Financing Newborn Screening

The program uses State General funds, the newborn screening fee ($10/infant) and Title V Maternal and Child Health Block grant funds to support access to treatment for the metabolic foods and formula. Title V Block grant funds support administrative aspects of the program (education, follow up, program management and quality assurance). The State General Fund appropriation has stayed the same since 1997, and the Title V Block grant appropriation to the State is below 1997 levels. The program continues to look for creative ways to make shrinking funds go further as costs increase.

Quality Assurance

In 2010 Quality Assurance Reports were sent to each birthing hospital and Children's Hospital in Omaha for the 1st and 4th quarters. An Office Suite change in State software prevented the reports from being produced in the intervening quarters. These reports included the individual hospital's quarterly measures on missing demographic information from the filter paper and a statewide comparison. The ability to produce the quality assurance reports of other measures such as turnaround time and unsatisfactory specimen rates for each hospital, continued to be unavailable due to the IT change. In addition, the publication "QI Hints" was sent out with each quality assurance report to the person(s) designated by the birthing hospital administrator.

Topics in 2010 included:

-UPS Internet tracking system for specimen shipments
-Regulation revisions affecting NICU admissions,
-Regulation revisions affecting how residual dried blood spots may be used
-Annual Report availability

Technical assistance visits were made to 7 hospitals in 2010. Responses on evaluations of the technical assistance visit consistently rated high on measures of usefulness of the visit.

NEWBORN SCREENING ADVISORY COMMITTEE

A huge debt of gratitude is owed to the dedicated members of the Newborn Screening Advisory Committee who commit their time and expertise to the Nebraska Newborn Screening Program. Much of Nebraska's success can be directly tied to their recommendations and guidance!

The Newborn Screening Advisory Committee (NBSAC) provided technical expertise and policy guidance to the Nebraska Newborn Screening Program. Members commit at least a half a day every three months to advise the state program. Representatives from PerkinElmer Genetics laboratory regularly provided input, presentations and proposals to the advisory committee. Several members provided extensive review and consultation beyond the committee meetings to help the program meet the recommendations of the larger committee.

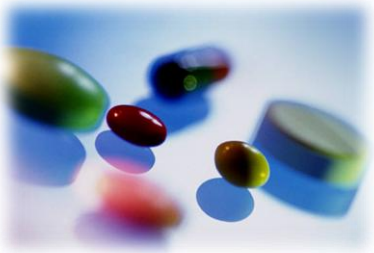
The members of the NBSAC in 2010 were:

- **CHAIR, James L. Harper**, M.D., *Pediatric Hematologist*, UNMC, Omaha
- **VICE-CHAIR Khalid Awad**, M.D., *Neonatologist*, Methodist Women's Hospital, Omaha
- **Lawrence Bausch**, M.D., *Neonatologist,* Lincoln
- **John Colombo,** M.D., *Pediatric Pulmonologist, Director,* Nebraska Cystic Fibrosis Center, UNMC, Omaha
- **Kevin Corley**, M.D., *Pediatric Endocrinologist*, Children's Hospital, Munroe/Meyer Institute for Genetics and Rehabilitation, UNMC, Omaha
- **Jeanne Egger,** *Parent***,** Hallam
- **David Gnarra,** M.D., *Pediatric Hematologist,* Children's Hospital, Omaha
- **Kathryn Heldt,** R.D., *Dietitian*, Children's Hospital Metabolic Clinic, Omaha

- **Mary Kisicki,** R.N., *Parent*, Papillion
- **Richard Lutz,** M.D., specialist in *Pediatric Genetics, Endocrinology, Metabolism,* Munroe/Meyer Institute for Genetics and Rehabilitation, UNMC, & Children's Hospital Omaha
- **Bev Morton,** *Parent,* Lincoln
- **Samuel Pirruccello**, M.D., *Pathologist*, Regional Pathology Services, UNMC, Omaha
- **Deborah Perry,** M.D., *Pathologist*, Pathology Center, Omaha
- **William Rizzo,** M.D.**,** specialist in *Pediatric Genetics, Endocrinology, Metabolism,* Munroe Meyer Institute for Genetics and Rehabilitation, UNMC, and Children's Hospital Omaha
- **Kathy Rossiter,** M.S.N, C.P.N.P., J.D., Omaha
- **Jill Skrabal,** R.D., *Dietitian*, Munroe Meyer Institute for Genetics and Rehabilitation, UNMC, and Children's Hospital Omaha
- **Corri Stearnes,** *Parent*, Omaha
- **William Swisher,** M.D., *Pediatrician,* Lincoln Pediatric Group, Lincoln
- **B.J. Wilson,** M.D., *Neonatologist/Perinatologist*, Saint Elizabeth Regional Medical Center, Lincoln

Assurance of Treatment and Management of Conditions

How Treatment and Management is Paid:

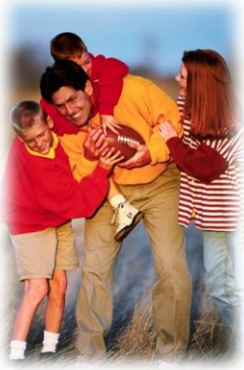


Part of the public health assurance role of Newborn Screening is ensuring treatment availability and access. Toward that end, the state program manages several contracts to ensure provision of otherwise prohibitively expensive formulas, foods, and services not always reimbursed by insurers. Approximately 65 patients received services through these contracts. (Some patients move out of state/new patients move in or are born/diagnosed with metabolic conditions).

Insurance usually covers medical treatments for some screened conditions such as prophylactic penicillin for patients with sickle cell disease, or synthetic thyroid hormone for patients with congenital primary hypothyroidism. However, many do not cover the metabolic formulas, and none cover the pharmaceutically manufactured foods required for PKU and other metabolic conditions screened. Therefore the biggest funding source supporting the metabolic foods and formulas was revenue generated from the $10 per infant screened fee (approximately $260,000 per year). The State General Fund appropriation of $42,000 also helped provide for these medically necessary formulas and foods and the associated nutritional counseling for patients identified with PKU or the other metabolic conditions identified on the tandem mass spectrometry screen. Title V Maternal and Child Health Block Grant funds then filled in the gaps for metabolic foods/formula and nutritional counseling. The Medically Handicapped Children's Program provides some assistance to eligible families with children who have a hemoglobinopathy such as sickle cell disease or those with cystic fibrosis.

Individuals affected with screened metabolic conditions can obtain the metabolic formula through the Nebraska Medical Center Adult Metabolic Clinic, or at the Children's Hospital Metabolic Clinic. Ongoing dietary consultation, pediatric metabolic specialty care and routine blood monitoring are also provided and necessary for proper management. Individuals can order the pharmaceutically manufactured foods from product lists provided by the 6 manufacturers/distributors that have contracts with the State. Families can order up to $2,000 of the pharmaceutically altered foods per year without having to pre-pay.

Nebraska's families:


In Federal Fiscal Year 2010, metabolic formula ordering and distribution and specialized nutritional counseling and monitoring were provided via a contract with the University of Nebraska Medical Center for $366,507. The individuals eligible for the metabolic foods utilized the pharmaceutically manufactured foods program, ordering foods with a value totaling $73,481.



Mike Rooney coordinates the day-to-day metabolic foods program helping families to understand the program and stay connected, and monitoring the vendors' compliance with the contracts. He provides a tracking log to families for their use in monitoring their orders and expenses and provides a mid-year spending report to each family. He also works closely with Jill Skrabal, RD to ensure timely contract amendments of appropriate metabolically altered food products as manufacturers continue to expand their offerings.

Sustaining the obligation to ensure access to treatment:

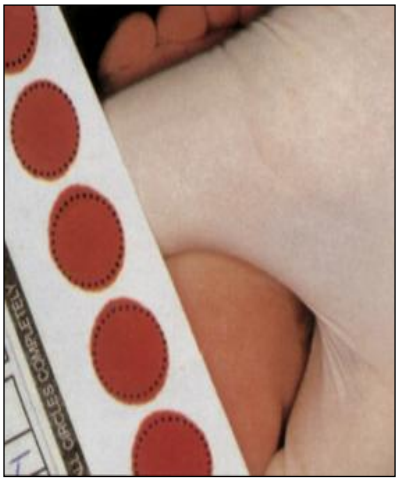
The number of children identified with conditions requiring special formula will always increase. The metabolic diets are required for life, so people do not "age-out" of the need for the special formulas or foods. State General Funds have remained flat and federal allocations to Nebraska of Maternal and Child Health Title V Block grant funds have been reduced or flat for several years. While a relative new drug is available for which about 40% of patients with PKU are expected to respond positively, these medications are expensive as well. Therefore the program continues to look for sustainable ways to continue to assure access to needed services for people who have these conditions.

Nebraska's Newborn Screening Fees

In 2010 the charge for newborn screening continued to be $38.50. The laboratory testing fee was $28.50 and the State fee (per statute and regulation) was $10.00 per infant screened. (State fee used only to help pay for treatment services). Hospital charges are separate and not regulated by the program. Based on the National NBS & Genetics Resource Center data, of the 47 states that charged a fee for newborn screening in 2010 only 5 were lower (FL, ID, LA, NC, TX).

PROCESS/OUTPUT DATA FOR 2010

SPECIMEN COLLECTION, HANDLING AND TRANSPORT



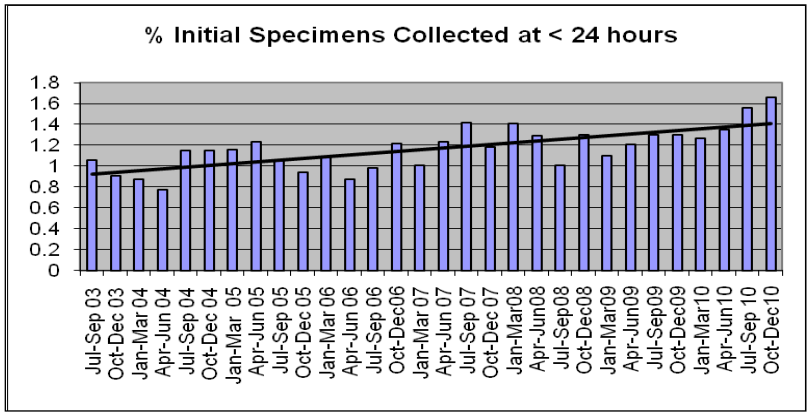
Age at Time of Specimen Collection (Initial Specimen) 2010

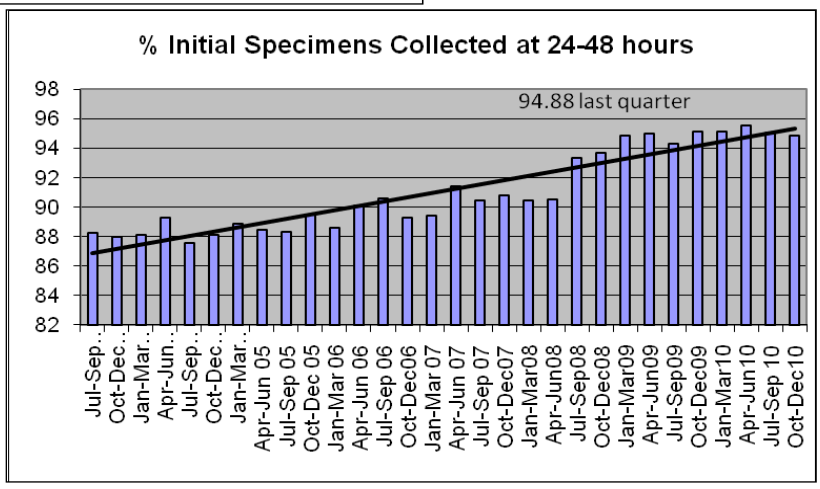
Age at time of collection	Number of births*	Percent of births
0-12 hours	243	.92
12-24 hours	144	.54
Collected day 2 (24-48 hours of age)	25195	1.46
Day 3	693	95.09
Day 4	72	2.62
Day 5	23	0.27
Day 6	21	0.09
Day 7	9	0.08
Over 7 days	94**	0.35
Time of collection unknown	1	0.00

***Number of births listed includes babies transferred in from other states that Nebraska's lab screened for the first time.**
****Initial specimens collected at greater than 7 days were from out-of-hospital births or hospital errors.**

Regulations require all specimens to be collected between 24-48 hours of birth, or prior to discharge, transfer or transfusion whichever comes first. Specimens collected past day two are at increased risk of a delayed diagnosis.

Hospitals improved from 94.75% in 2009 to 94.88% of births screened at the correct time (between 24-48 hours of birth) in 2010.


% Initial Specimens Collected at < 24 hours
1.8
1.6
1.4
1.2
1
0.8
0.6
0.4
0.2
0
Jul-Sep 03
Oct-Dec 03
Jan-Mar 04
Apr-Jun 04
Jul-Sep 04
Oct-Dec 04
Jan-Mar 05
Apr-Jun 05
Jul-Sep 05
Oct-Dec 05
Jan-Mar 06
Apr-Jun 06
Jul-Sep 06
Oct-Dec06
Jan-Mar 07
Apr-Jun 07
Jul-Sep 07
Oct-Dec 07
Jan-Mar08
Apr-Jun08
Jul-Sep08
Oct-Dec08
Jan-Mar09
Apr-Jun09
Jul-Sep09
Oct-Dec09
Jan-Mar10
Apr-Jun10
Jul-Sep 10
Oct-Dec10


% Initial Specimens Collected at 24-48 hours
94.88 last quarter
98
96
94
92
90
88
86
84
82
Jul-Sep..
Oct-Dec..
Jan-Mar..
Apr-Jun..
Jul-Sep..
Oct-Dec..
Jan-Mar..
Apr-Jun 05
Jul-Sep 05
Oct-Dec 05
Jan-Mar 06
Apr-Jun 06
Jul-Sep 06
Oct-Dec06
Jan-Mar 07
Apr-Jun 07
Jul-Sep 07
Oct-Dec07
Jan-Mar08
Apr-Jun08
Jul-Sep08
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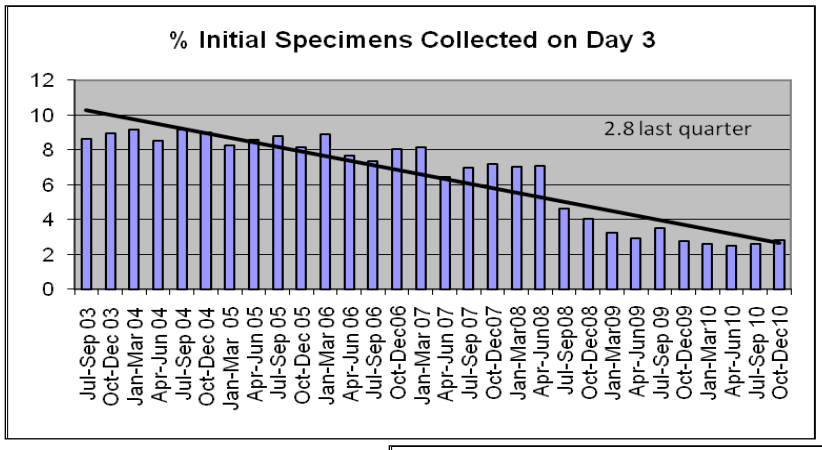
Oct-Dec09
Jan-Mar10
Apr-Jun10
Jul-Sep 10
Oct-Dec10


% Initial Specimens Collected on Day 3
2.8 last quarter
12
10
8
6
4
2
0
Jul-Sep 03
Oct-Dec 03
Jan-Mar 04
Apr-Jun 04
Jul-Sep 04
Oct-Dec 04
Jan-Mar 05
Apr-Jun 05
Jul-Sep 05
Oct-Dec 05
Jan-Mar 06
Apr-Jun 06
Jul-Sep 06
Oct-Dec06
Jan-Mar 07
Apr-Jun 07
Jul-Sep 07
Oct-Dec07
Jan-Mar08
Apr-Jun08
Jul-Sep08
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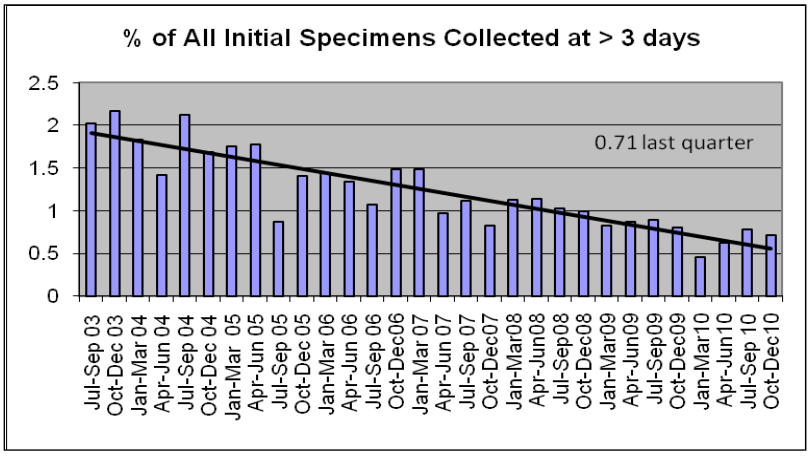
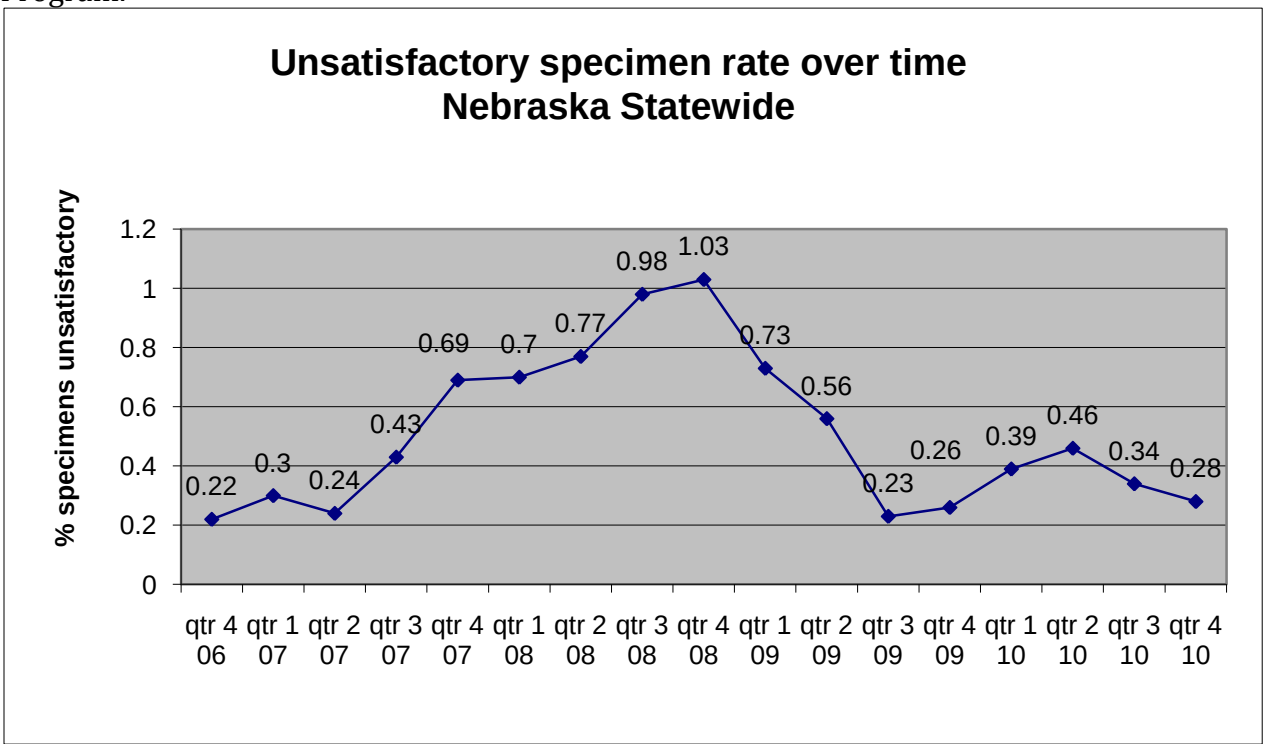
Jul-Sep 09
Oct-Dec09
Jan-Mar10
Apr-Jun10
Jul-Sep 10
Oct-Dec10


% of All Initial Specimens Collected at > 3 days
0.71 last quarter
2.5
2
1.5
1
0.5
0
Jul-Sep 03
Oct-Dec 03
Jan-Mar 04
Apr-Jun 04
Jul-Sep 04
Oct-Dec 04
Jan-Mar 05
Apr-Jun 05
Jul-Sep 05
Oct-Dec 05
Jan-Mar 06
Apr-Jun 06
Jul-Sep 06
Oct-Dec06
Jan-Mar 07
Apr-Jun 07
Jul-Sep 07
Oct-Dec07
Jan-Mar08
Apr-Jun08
Jul-Sep08
Oct-Dec08
Jan-Mar09
Apr-Jun09
Jul-Sep09
Oct-Dec09
Jan-Mar10
Apr-Jun10
Jul-Sep 10
Oct-Dec10

Unsatisfactory Specimens for 2010

Although Nebraska's unsatisfactory specimen rate was increasing, it was still among the lowest of unsatisfactory rates in the U.S. However, because every unsatisfactory specimen requires the baby to have another specimen collected, and creates the potential for a delayed diagnosis, the program takes this issue very seriously. After two and a half years of QA and aggressive educational effort, State averages finally returned to the good performance levels routinely seen prior to mid 2007.

The effort to reduce unsatisfactory specimens is valuable because they can be costly on many levels. Repeat screens must be done requiring effort on the part of newborn screening follow up, hospital, screening lab and physician office personnel, plus the effort and inconvenience to families to have to return to the hospital for the repeat heel stick procedure on their infant. Although the screening laboratory does not charge for requested repeat specimens, hospital phlebotomy charges may apply. Maintaining low unsatisfactory specimen rates is a high priority goal of the Nebraska Newborn Screening Program.



The art and science of correctly collecting and handling dried blood spots on filter paper requires trained health care professionals, who consistently follow the Clinical and Laboratory Standards Institute procedures for specimen collection. <u>Every</u> unsatisfactory specimen must be repeated, to ensure sufficiently reliable screening results.

Drawn Early Specimens for 2010
(less than 24 hours of age)

YEAR 2010
SUMMARY OF DRAWN EARLY DATA

January 1, 2009 – December 31, 2009

#DE's per month:

Jan	21	12 transferred, 2 pre-transfusion, 7 reason unknown	(0 expired)
Feb	17	11 transferred, 1 pre-transfusion, 5 reason unknown	(1 expired)
Mar	20	8 transferred, 2 pre-transfusion, 10 reason unknown	(0 expired)
Apr	24	17 transferred, 5 pre-transfusion, 2 reason unknown	(1 expired)
May	24	14 transferred, 1 pre-transfusion, 9 reason unknown	(1 expired)
June	24	14 transferred, 2 pre-transfusion, 8 reason unknown	(2 expired)
July	34	22 transferred, 5 pre-transfusion, 1 early discharge, 6 unknown,	(0 expired)
Aug	27	21 transferred, 5 pre-transfusion, 1 reason known	(0 expired)
Sep	34	20 transferred, 4 pre-transfusion,10 reason unknown	(0 expired)
Oct	28	20 transferred, 1 pre-transfusion, 1 early discharge, 6 unknown,	(0 expired)
Nov	28	20 transferred, 2 pre-transfusion, 6 reason unknown	(3 expired)
Dec	21	12 transferred, 3 pre-transfusion, 6 reason unknown	(2 expired)

TOTAL: 304 + 8 expired

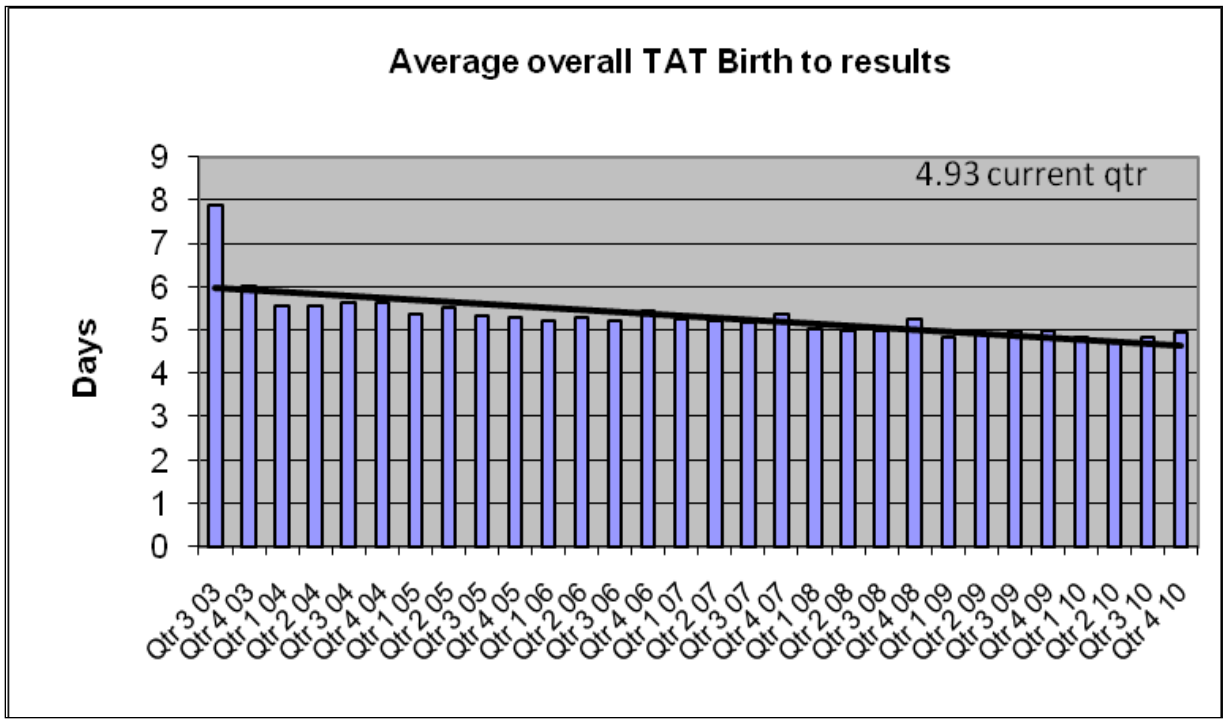
****Note:** There were additional infants that were reported as drawn early and upon notification to the birthing facility it was reported and documented that there was a reporting error made.

Specimen Turnaround Time

Regular monitoring of turnaround time between birth and reporting of results of the initial specimen is an important indicator for how well the newborn screening system is functioning.

On the positive side, overall turnaround times continued to decline in 2010 thanks to in-lab efforts at PerkinElmer Genetics, and hospital personnel responding to the quality assurance reports when turnaround times for collection were above the benchmark/ average of 1.5 days of age.

The Newborn Screening Advisory Committee reviews the quarterly results of average times from: birth to collection, collection to receipt in the lab, in-lab turnaround time, and overall turnaround time from birth to reporting out of results as in the following graphic in which the last quarter of 2010 ended up with average of 4.93 days.



LABORATORY TESTING DATA

PerkinElmer Genetics Inc. Laboratory uses several instruments to complete the testing. While tandem mass spectrometry provides the screening for 20 of the required conditions, other methods are used for the other 8.

Presumptive Positive, Inconclusive, & Confirmed Positive Numbers & Rates

Screening Rates

Screening programs by their very nature are designed to find those at higher risk of a disease in order to facilitate their diagnosis and treatment to prevent morbidity and mortality. Screening tests were never designed to be diagnostic and so a small percent of screen results will be positive that upon repeat or confirmation are found to be normal. Nebraska and programs across the country strive to minimize the number of newborns that require repeat or confirmatory testing (presumptive positive), and maximize the probability of identifying those affected. Nebraska continued to sustain a relatively low false positive rate for every condition screened.

Most of the babies requiring any follow up for abnormal results in Nebraska require only a repeat dried blood spot specimen which usually has a normal result.

- When a screening result is reported out as "inconclusive" the recommended follow up is a repeat dried blood spot specimen. (Most of these will be normal on repeat).
- When a screening result is reported out as "presumptive positive," the follow up is treated more urgently and usually a confirmatory test by a different method or on a different kind of specimen (serum, whole blood, urine etc.) is necessary.

Often the results are abnormal primarily because the baby was premature, sick, low birth weight, or receiving special treatment such as parenteral nutrition which can interfere with newborn screening results. These babies account for a disproportionate amount of the follow up needed. However this is not an argument to delay screening on these babies as they are at equal or possibly higher risk of having one of the screened conditions.

Condition Screened **2010 Data**	**# Screened**	**# Presumptive Positive or inconclusive on initial screen**	**Presumptive Positive Rate**	**# lost to follow up**	**# confirmed Positive/ Diagnosed (classical or partial w tx/)**
Biotinidase deficiency	26,176	18	0.06%	0	1
Congenital Adrenal Hyperplasia	26,176	38 (25 of these were inconclusive)	0.14%	4 (2 of these expired)	2
Congenital Primary Hypothyroidism	26,176	135 (70 of these were inconclusive)	0.51%	1 (expired)	11
Cystic Fibrosis	26,176	125 (123 of these were inconclusive)	0.47%	7 (expired)	4
Galactosemia	26,176	8	0.03%	0	1 (duarte)
ASA (see*)	26,176	1	.003%	0	1
Hypermethioininemia (see*)	26,176	89	0.3%	0	1
LCHAD (see*)	26,176	1	.003%	0	1
MCAD (see*)	26,176	2	.007%	0	1
MMA (see*)	26,176	31	0.1%	0	3
PKU (see*)	26,176	4	.01%	0	2
SCAD (see*)	26,176	4	.01%	0	1
Sickle Cell Disease & other clinically significant hgbs	26,176	9	0.03%	Dx unknown	9
All other abnormal Hgb's (carriers/variants)	26,176	467	1.78%	215 dx unknown, but 157 of those had confirmatory testing	

* babies lost to follow up expired before repeat or confirmatory testing could be completed

** 113 abnormalities from the MS/MS testing were elevations of multiple amino acids consistent with babies who were receiving parenteral nutrition

Mean Averages of Laboratory Test Measures

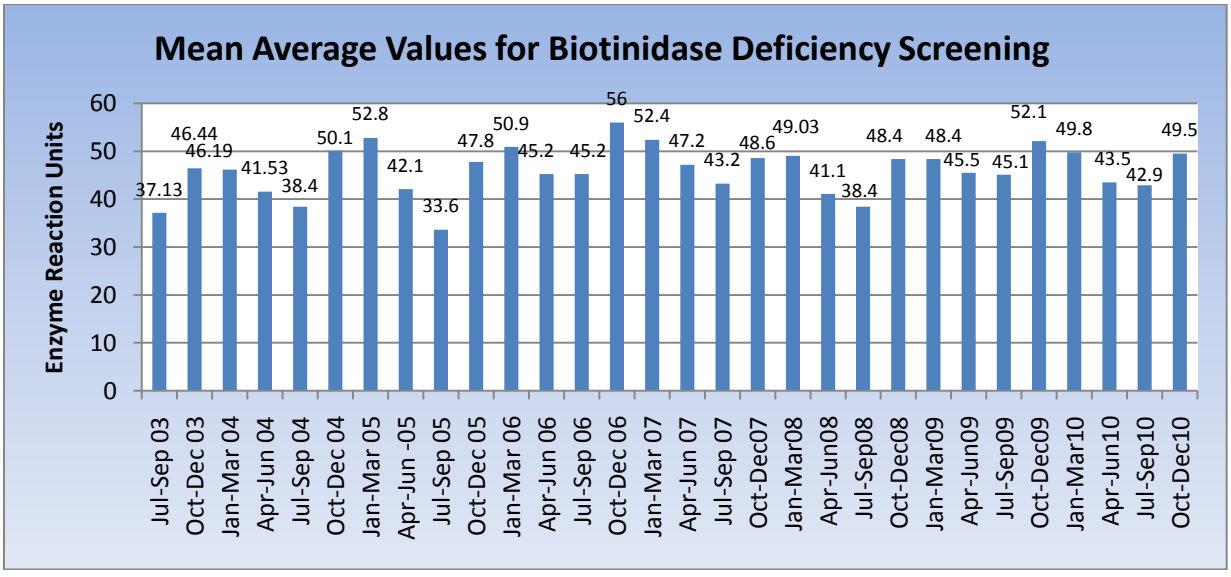
The program continues to provide lab testing data to the Newborn Screening Advisory Committee to monitor ongoing quality. The following tables depict the quarterly mean averages for biotinidase measures, 17-OHP for congenital adrenal hyperplasia, Immunoreactive trypsinogen for CF, GALT, and total galactose used to screen for Galactosemia. Access to data for mean averages for the amino acids and acylcarnitines used to screen for the fatty acid, amino acid and organic acid disorders are not available from the Tandem Mass Spectrometry results from the screening laboratory. The T4 and TSH results are not included because some results were beyond the linearity of the assay prior to 2010 and would affect the accuracy of this data. These means can tell us something about stability of the assay, reagents etc. over time.

Health care providers familiar with the mean averages might feel more comfortable explaining the "relative risk" to parents of newborns with positive screening results, by comparing how far out of range the result is from the mean average, and from the normal expected range.

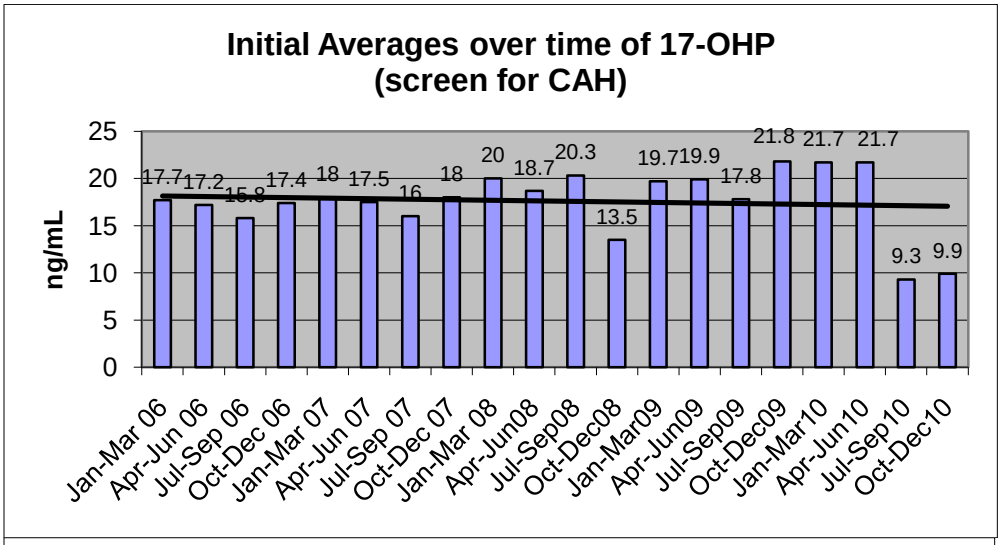
Expected seasonal differences can be seen each summer when heat exposure may impact the mean average enzyme levels detected in screening for biotinidase deficiency.

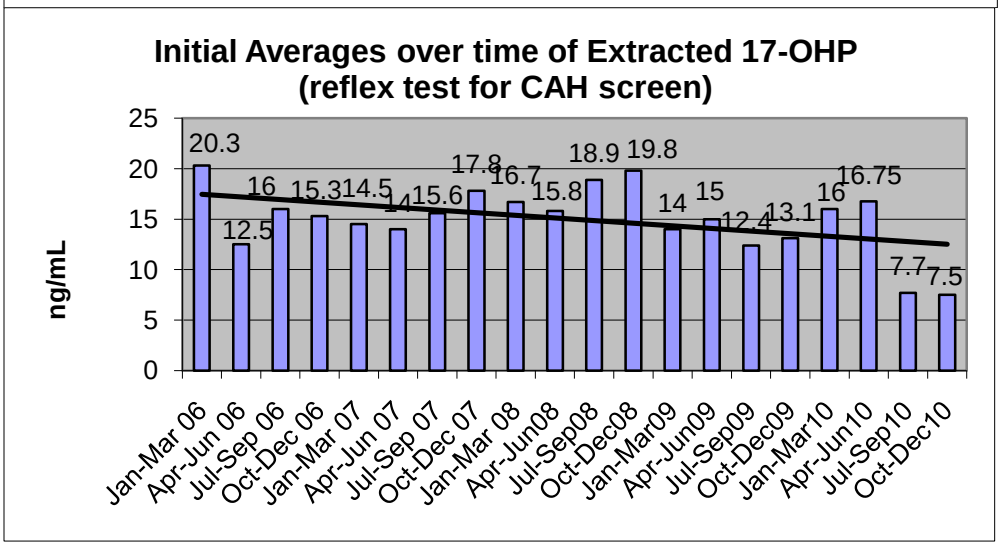
The Nebraska Newborn Screening Program sends a reminder each spring to hospital laboratories about specific practices to follow that will minimize the risk of specimens becoming heat denatured. This is intended to avoid the associated increase in the number of rejected specimens.

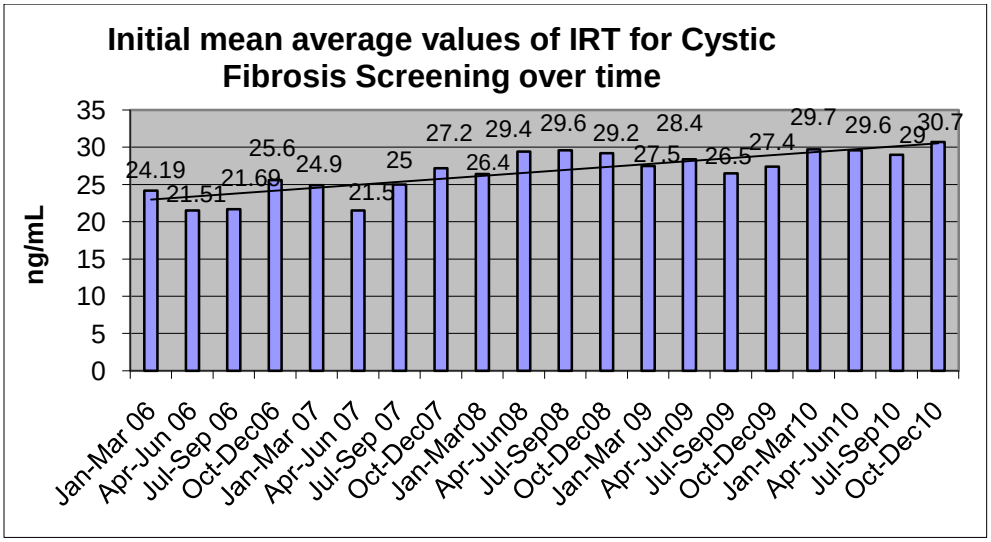
In 2010, only 2 specimens were rejected and needed repeats to re-test enzyme assays used to screen for conditions such as biotinidase deficiency and galactosemia because the initial specimen had been exposed to heat/humidity. This continued the improved trend from 29 needing repeats in 2008, and 6 in 2009. That is an outstanding testament to the care hospitals take in handling these specimens!



Reflex testing of abnormal CAH screens using an extracted 17-OHP reduces the number of positive screens reported and needing confirmatory testing. The extracted 17-OHP is thought to minimize the effects of interfering substances.





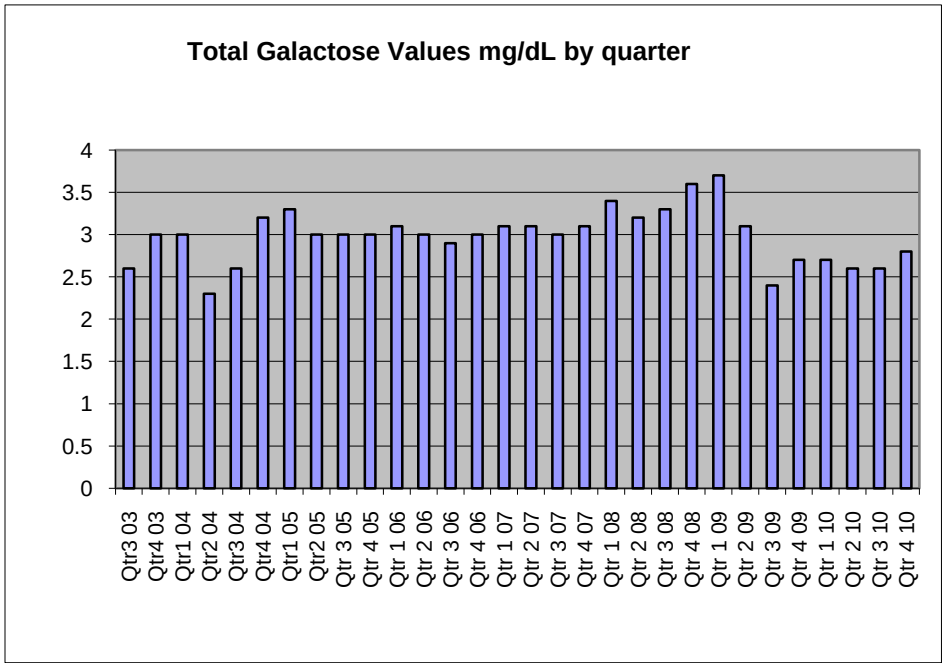


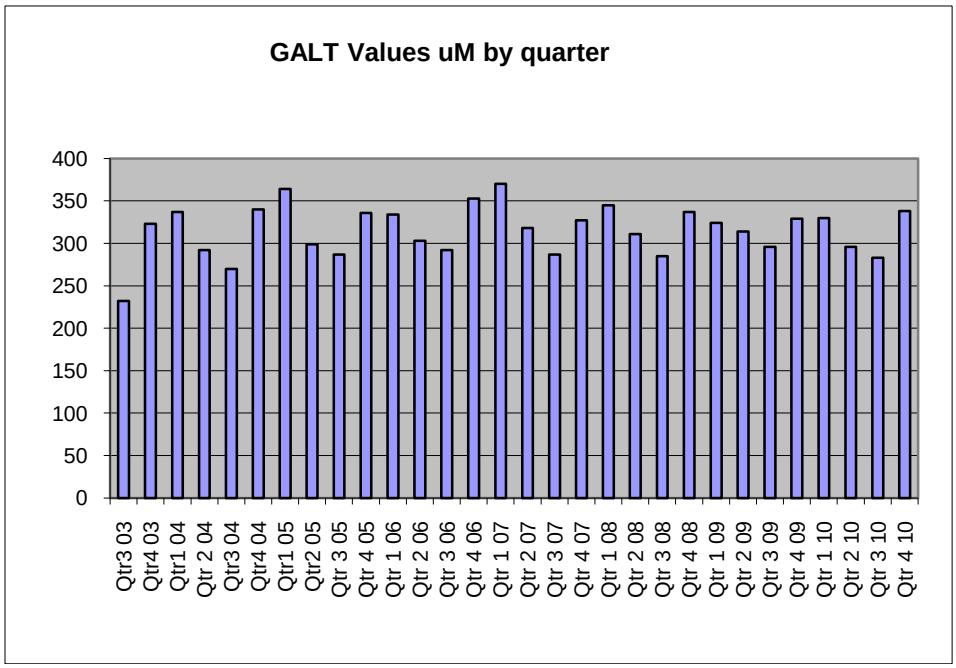
IRTs greater than 90 reflexed to test for ΔF508 the mutation most commonly associated with classical cystic fibrosis. (Those with one copy of the ΔF508) reflexed to testing for additional mutations on the Luminex 39 mutation panel).

By looking at both elevations of galactose, and decreases in the enzyme activity of galactose phosphate uridyl transferase the laboratory can report with greater precision, those newborns at risk for classical Galactosemia . These need immediate metabolic consultation/ referral and testing, vs. those whose findings are more consistent with a milder but potentially clinically significant form of Galactosemia.

Having this information can mean providing more parents with a bit more peace of mind since most will need only a repeat screen, vs. full confirmatory testing.

This also can translate into cost savings because doing a "requested repeat" screen at no charge may be all that is initially recommended, vs. more expensive confirmatory testing.





NEWBORN SCREENING DATA

	2001	2002	2003	2004	2005	2006	2007	2008	2009	2010
Total Births	25,109	25,515	26,067	26,443	26,349	26,898	27,107	27,094	27,199	26,243
Births Screened	25,043 99.7%	25,478 99.85%	26,008 99.77%	26,391	26,288	26,819	27,013	27,021	27,131	26,176
Total Births Lost to Follow up	2 + (64 not screened as expired @ < 48 hours)	5 + (32 not screened as expired @ < 48 hours)	5 + (54 not screened as expired @ < 48 hours)	2 + (50 not screened as expired @ < 48 hours)	0 + (61 not screened as expired @ < 48 hours)	2 + (79 not screened as expired @< 48 hours)	(94 not screened as expired @ < 48 hours)	1 (+ 73 not screened as expired @ < 48 hours)	3 (67 not screened as expired @<48 hrs, 1 discharged w/o screen & out of state)	12 (10 of these expired @ < 48 hrs. of age
Total Births PP**	432	456	415	499	503	537	511	553	99 + 912 (see foot note)	949
Home Births	93	99	70	60	55	69	80	86	99	96
Home Births Screened	88	95	65	60	54	69	78	85	97	96
Home Births Lost to follow up[1]	2 + (3 expired)	2 + (2 expired)	3 + (2 expired)	0	0 + (1 expired)	0	2 (both expired)	1 (expired)	2 (expired)	0

*Began match with death records beginning in calendar year 2000, to more accurately report #s actually screened.
** PP = Presumptive Positive. Includes all initial screen results requiring either a repeat dried blood spot or another confirmatory specimen and test.

Note: In 2009 Nebraska began counting the number of screens presumptive positive as those requiring confirmatory testing because of a more substantial out-of range result (99). The 912 out of range results listed required only repeat testing of a dried blood spot filter paper. The number requiring confirmatory tests in 2010 included 111(metabolic/endocrine/CF). Positive hemoglobins received confirmatory testing for 417 of 475 recommended.

Biotinidase Deficiency	2001	2002	2003	2004	2005	2006	2007	2008	2009	2010
Presumptive Positive	4	3	4	34*	78	14	5	4	5	1
Inconclusive							10	25	17	17
Confirmed Negative	1	1	0	29	71	9	11	23	17	17
Confirmed Positive Profound	0	2	1	0	1	0	0	1	0	0
Confirmed Positive (Partial no tx)	0	0	0	0	0	0	0	0	0	0
Confirmed Positive (Partial tx)	3	0	3	6	5	4	4	3	5	1
Lost to follow up	0	0	0	0	1**	1**	0	2***	0	0

*Screening protocols identified most of these as "inconclusive," for which repeat screening rather than confirmatory testing, ruled out the condition.
** lost to follow up as newborn expired

Congenital Adrenal Hyperplasia	2001	2002	2003	2004	2005	2006	2007	2008	2009	2010
Presumptive Positive	N/A	N/A	N/A	N/A	N/A	10	3	17	7	12
Inconclusive							18	22	25	26
Confirmed/ repeated Negative	N/A	N/A	N/A	N/A	N/A	9	17	36	31	32
Confirmed Positive	N/A	N/A	N/A	N/A	N/A	1	1	1	1	2
Confirmatory or Repeat Lost to follow up	N/A	N/A	N/A	N/A	N/A	0	3*	2*	0	4*

* expired before repeat or confirmatory testing could be done.

Congenital Primary Hypothyroidism	2001	2002	2003	2004	2005	2006	2007	2008	2009	2010
Presumptive Positive	115	129	89	63	58	51	39	57	56	65
Inconclusive (drawn early but low T4/high TSH							20	52	48	71
Confirmed Or repeated	105	113	75	55	48	41	43	96	88	124

Negative										
Confirmed Positive	7	15	11	8	9	10	16	12	15	11
Confirmatory or Repeat Lost to follow up	3*	1*	3*	0	1*	0	3*	1	1*	1*

*Lost to follow up as newborn expired.

Data for Cystic Fibrosis and Hemoglobinopathies are presented in a different format because screening for CF is inherently more complex, and diagnosis for hemoglobinopathies can be more protracted and complex. Although the goal is to detect clinically affected newborns to initiate early treatment and prevent infant mortality and morbidity, the screening test can detect some carriers or people who have the trait for these conditions.

***Cystic Fibrosis*:**	**Year**	**2006**	**2007**	**2008**	**2009**	**2010**
Total Screened Positive		8	4	9	4	1
Of those:	Confirmed CF	4	8	9	3	1
	Confirmed Atypical CF	0	0	0	0	0
	CRMS (CF related metabolic syndrome)	0	0	0	1	0
Total Screened Inconclusive		62	54	53	50	122*
Of those:	Confirmed CF	3	2	5	0	1
	Confirmed Atypical CF	0	2	9	0	0
	CF Related Metabolic Syndrome				4	0
	Confirmed Carriers	12	10	6	9	10
	Found to be within normal limits on repeat	35	46	30	32	95
	Expired before confirmation could be done	4	1	6	2	9
	Lost to follow up	0	0	0	1	0
	Pending	0	1	0		2
Total with Meconium Ileus or Bowel Obstruction		4	13	1	8	3
Of those:	Confirmed CF	5	1	1	2	2
	Found to be within normal limits	7	3	0	6	1
	Pending diagnosis	1	0	0	0	0

* includes 3 that were within normal limits on follow-up with sweat chlorides

Galactosemia	**2001**	**2002**	**2003**	**2004**	**2005**	**2006**	**2007**	**2008**	**2009**	**2010**
Presumptive Positive	15	5	3	9	1	8	0	0	2	1
Inconclusive repeat rec'd	n/a	n/a	n/a	n/a	n/a	n/a	9	9	10	7
Confirmed / repeated Negative	9	5	0	6	1	8	8	9	12	7
Confirmed Positive (Classical)	0	0	1	0	0	0	0	0	0	0
Confirmed Positive, Duarte (not treated)	0	0	1	0	0	0	0	0	0	0

Confirmed Positive, Duarte (treated)	6 Duarte Mixed Htrzgt.	0	1	3	0	0	1	0	0	1

Hemoglobinopathy Follow up Changes:
Since 2006 follow up procedures included sending a reminder letter to the baby's physician before the 6 month checkup when the initial confirmatory report indicated a possible alpha, beta or gamma chain variant or combination in the heterozygous state. These typically require additional blood work to diagnose, which previously was not usually reported back to the program. Often these were hemoglobin patterns that had Bart's present on the initial screen and the concern was a possible alpha Thalassemia. This has resulted in a significant increase of diagnosed and closed cases. Ultimately the goal is to provide families with better information about their child's hemoglobinopathy.

Abbreviation Key (Likely diagnosis associated with screening results)

FS:	Sickle Cell Disease	FAS:	Sickle Cell Trait
FC:	Hemoglobin C Disease	FAC:	Hemoglobin C Trait
FSC:	Sickle Hemoglobin C Disease	FAD:	Hemoglobin D Trait
FE:	Hemoglobin E Disease	FAE:	Hemoglobin E Trait
FSA:	Sickle Beta Thalassemia	FAV:	Hemoglobin Trait - unknown variant
HPFH: Hereditary Persistence Fetal Hemoglobin		FA + Barts: Possible alpha Thalassemia	

Clinically Significant Hemoglobinopathies Confirmed Positive:

	2001	2002	2003	2004	2005	2006	2007	2008	2009	2010
FS	4	4	5		1	3		3	1	5
FC				1	1		1*		1	2
FSC	2	2		1	1	2	1	1	5	1
FE						1			1	
Sickle Beta Thal							1		3	1 C-beta thal
Alpha Thal Major	1 (4-gene deletion)									
Beta Thal Major							1		1	
HPFH					1					
FAE + possible Beta Thal						6				
FAS + possible Beta Thal						11				

FAS + Alpha Thal						9				
FAC + Alpha Thal						3				

Dx. = Sickle Hemoglobin C Disease or Hemoglobin C beta Thalassemia

Other Hemoglobinopathies Confirmed Positive in 2010:

149 Sickle Cell Trait
5 Hgb. D Trait
7 Alpha Thal Trait
38 Hgb. C Trait
29 Trait + other
12 miscellaneous traits
10 Hgb. E Trait
1 Alpha Thal silent carrier
1 sickle trait + Alpha Thal trait

215 Confirmatory diagnosis unknown. (Confirmatory testing done for 157 of those, but no final diagnosis. None known to be clinically significant.)

MCAD *	2001	2002	2003	2004	2005	2006	2007	2008	2009	2010
Screened Positive	N/A	3*	3	5	10	5	0	4	3	2
Screened inconclusive (repeat only)**	N/A	N/A	N/A	N/A	N/A	N/A	2	2	8	0
Confirmed Negative or Repeated normal	N/A	2	3	1	7	5	2	2	8	1
Confirmed Positive	N/A	1	0	4	3	0	0	4	3	1

*Mandatory screening for MCAD began 7/01/2002. Prior to that about 34% of newborns were voluntarily screened in Nebraska in 2000 and 2001.
**Inconclusive screen: Abnormal screen result requiring only a repeat screen, not confirmatory testing.

Phenylketonuria (PKU)	2001	2002	2003	2004	2005	2006	2007	2008	2009	2010
Presumptive Positive	4	3	7**	7	3	6	0	0	4	4
Screened Inconclusive (repeat only)***	N/A	N/A	N/A	N/A	N/A	N/A	2	0	1	0
Confirmed Negative	2	1	1	1	1	1	2	0	1	0
Confirmed Positive Classical PKU	1	1	2	1	2	0	0	0	3	2
Confirmed Positive Hyperphe	1	1	3	5 (3 of these tx'd)	0	5 (4 of these treated)	0	0	1	2

****2000 and 2003**: One each year for whom confirmatory testing was not done as the babies expired
***Inconclusive screen: Abnormal screen result requiring only repeat screen, not confirmatory testing.

Tandem Mass Spectrometry Screening Results (MS/MS)

Initial findings	# abnormal On screen	# confirmed negative	# pending or lost to follow up	# confirmed positive
Methionine	76 + 4 on repeat	79		1 hypermethioninemia
Several Amino Acids	107 + 5 on repeat	112		
A generalized elevation of short-chain & medium-chain acylcarnitines	3 + 2 on repeat	5		
Propionylcarnitine (C3)	25 + 1 on repeat	26		
C3 & C3/C2 & C3/C16	18	15		3 methylmalonic academia
Arginine	1			1 argininosuccinic aciduria
C4 Butyrylcarnitine	3	3		1 short chain acyl CoA dehydrogenase deficiency
C4 Butyrylcarnitine and other indices such as the relation ratio of C4 to Propionylcarnitine (C3)	1	1		
C5	2	2		
C5 & C4	1	1		
C5OH	2	2		
C8 Octanoylcarnitine	2	1		1 medium chain acyl CoA dehydrogenase deficiency
Methionine & Phenylalanine	2	2		
Methionine & Tyrosine	12	12		
Tetradecenoylcarnitine (C14:1) to C16 & other long chain acylcarnitines	1			1 long chain acyl CoA dehydrogenase deficiency
3-Hydroxyisovalerylcarnitine (C5OH)	2	2		
Tyrosine	23	16		7 transient tyrosinemia
2010 Totals (NE Infants)	**293**	**278**		**15**

*Lost to follow up designated when the patient/parent can no longer be found and there is no medical home, or they have moved out of state to an unknown location.
**The vast majority of abnormal screens from MS/MS require only a repeat screen to rule out the condition. Confirmatory testing is recommended in a small percentage of cases where the concentration of analytes are "significantly" abnormal, or concentrations of analytes increase on repeat screens.
***Total babies less than # of abnormal screens as some that had more than one abnormal screen, e.g. methionine on initial screening, and multiple amino acids on a repeat screen.



Intervention Data

Intervention data is one of the most important measures for determining how well we are doing as a system to ensure timely treatment of affected infants.

Several factors can conspire to create delays in treatment, so speed and persistence in follow up are essential. Some examples of these factors include babies with prolonged treatment in NICUs, parental resistance to confirmatory testing, problems in locating parents because contact information provided to the hospital or recorded on the filter paper collection cards was incorrect or no longer accurate.

Condition & number of babies diagnosed	Average age at intervention/tx.	Range in ages at intervention/tx.
1 Argininosuccinic Aciduria	44 days	44 days
1 Biotinidase Deficiency	12	12
2 Congenital Adrenal Hyperplasia	5	4-6
11 Congenital Primary Hypothyroidism	10	6-29
4 Cystic Fibrosis	27	7-62
1 Duarte Galactosemia	13	13
1 Long Chain Acyl CoA Dehydrogenase def	4	4
1 Medium Chain Acyl CoA Dehydrog. Def	6	6
3 Methylmalonic Acidemia	14	5-27
1 Short Chain Acyl CoA Dehydrogenase def	14	14
1 Hypermethinioninemia (not treated)	Confirmed @ 16	
2 Hypephenylalaninemia (1 not treated)	7	7
2 Phenylketonuria	7	7
2 Hgb. C Disease	54.5	42-67
5 Sickle Cell disease	26	18-39
6 Transient Tyrosinemia (3 treated)	11.33	6-14
1 Hgb SC Disease	22	22
1 C-Beta Thalassemia	Unknown, child left State	

NEBRASKA EARLY HEARING DETECTION AND INTERVENTION ANNUAL REPORT - 2010

Introduction

Significant hearing loss is one of the most common birth conditions with an estimated incidence of one to three per thousand live births. Before newborn hearing screening, many hearing losses were not diagnosed until 2 ½ to 3 years of age. Left undetected, hearing loss in infants can negatively impact speech and language acquisition, academic achievement, and social and emotional development. If detected soon after birth, the negative impacts can be diminished and even eliminated through early intervention.

In 2000, the Infant Hearing Act established newborn hearing screening in Nebraska. The Nebraska Early Hearing Detection and Intervention (NE-EHDI) Program strives to fulfill the following four purposes of the Infant Hearing Act (Neb. Rev. Stat. §71-4735):

- To provide early detection of hearing loss in newborns at the birthing facility, or as soon after birth as possible for those children born outside of a birthing facility;
- to enable these children and their families and other caregivers to obtain needed multidisciplinary evaluation, treatment, and intervention services at the earliest opportunity;
- to prevent or mitigate the developmental delays and academic failures associated with late detection of hearing loss; and
- to provide the state with the information necessary to effectively plan, establish, and evaluate a comprehensive system for the identification of newborns and infants who have a hearing loss.

The Act required birthing facilities to educate parents about newborn hearing screening, to include hearing screening as part of the standard of care, and to establish a mechanism for compliance review by December 2003. The Act also required that regulations be promulgated to mandate newborn hearing screening if less than 95% of newborns in the state received a hearing screening.

Newborn hearing screening requires objective physiologic measures to detect hearing loss in newborns and young infants. There are two basic techniques available to screen newborns for hearing loss. Both are easily performed on newborns and are non-invasive measures of physiologic activity that underlie normal auditory functioning.

The most frequently used screening technique is measurement of otoacoustic emissions, or OAEs. A miniature earphone and microphone are placed in the newborn's ear canal, low intensity sounds are presented, and responses produced by the inner ear are measured. The second screening technique, Auditory Brainstem Response, or ABR, uses small electrodes to detect certain brainwaves in response to sounds that are presented by a

miniature earphone. For both methods, the response of each ear is measured. OAE and ABR are both reliable and accurate. Screening can occur as early as 12 hours of age, preferably with the newborn sleeping, and averages from five to 20 minutes to complete.

If a response is not detected for one or both ears, the result is a "refer" (did not pass). A "refer" to the screening test indicates that a hearing loss *may* exist but there are also other factors that may have contributed. A "refer" does indicate that a second screening is necessary to determine if the other factors, such as vernix in the ear canal, fluid in the middle ear cavity, movement, equipment failures, or inexperience of the tester, contributed to the initial result. A "refer" on the second screening indicates the need for a diagnostic audiologic evaluation to confirm or rule out a hearing loss and, if hearing loss is present, to identify the type and degree of the loss and to begin intervention services.

Each birthing facility has established a newborn hearing screening protocol that identifies how the screening will be administered, the recording and reporting procedures, how "refers" will be handled, i.e., re-screen as an inpatient with the same or different screening technique or re-screen as an outpatient, and quality assurance measures.

Newborn Hearing Screening Data Reported for 2010

Birthing Facility Screening Programs

The number of birthing facilities conducting newborn hearing screening increased rapidly from 2000 when only 11 hospitals were conducting either targeted or universal newborn hearing screening (see Table 1). Since 2003, 100% of the birthing facilities in Nebraska have been conducting hearing screenings, consistent with the Neb. Rev. Stat. §71-4742 requirement that a hearing screening test be included as part of the standard of care for newborns. Fifty-eight (58) of the 59 birthing hospitals conduct the hearing screening during the birth admission and one conducts the screening on an outpatient basis following discharge.

Birthing Facilities Conducting Newborn Hearing Screenings (2000-2010)

Year	Number of Birthing Facilities in Nebraska	Number Conducting Newborn Hearing Screening	Percentage Conducting Newborn Hearing Screening
2000	69	11	16%
2001	69	24	35%
2002	69	57	83%
2003	67	67	100%
2004	67	67	100%
2005	65	65	100%
2006	63	63	100%
2007	63	63	100%
2008	61	61	100%
2009	58	58	100%
2010	59	59	100%

Table 1

Annual Birthing Facility Reports

Birthing facilities are required to annually report specific information about their newborn hearing screening programs to the Department of Health and Human Services (Neb. Rev. Stat. §71-4739). The ERS-II data system, an integrated module of the State's Vital Records system, automatically calculates these figures for each birthing facility.

Birthing Facility Reports of Data (2010)

Number of newborns (hospital births with transfers of non-hospital births)	26,166
Number of newborns available for hospital inpatient screenings (excluding expired newborns)	26,066
Number of newborns who received a hearing screening during birth admission	25,925*
Number of newborns who passed a hearing screening during birth admission, if administered	24,796*
Number of newborns who did not pass a hearing screening during birth admission, if administered	1,129*
Number of newborns recommended for monitoring, intervention, follow up care	649

*Includes babies transferred to Children's Hospital & Medical Center, non-birthing facility *Table 2*

Parent Education

Recommending a hearing screening test has been operationally defined as educating parents about newborn hearing screening, hearing loss, and normal communication development as required by Neb. Rev. Stat. §71-4740. The NE-EHDI Program provides print and video educational materials free of charge to hospitals to help fulfill this requirement. Print materials are available in 10 languages. Birthing facilities reported educating almost all parents (25,652 or 98%) about newborn hearing screening, hearing loss and normal speech and language development in 2010. Neb. Rev. Stat. §71-4740 requires the Department of Health and Human Services to educate parents of newborns who are not born in a birthing facility about the importance of newborn hearing screening and to provide information to assist them in having the screening performed within one month after the child's birth. There were 94 babies recorded as having been born out-of-hospital in 2010. Parent education material is sent to the parents of the babies who were not admitted to a hospital immediately following birth.

Newborns Receiving a Hearing Screening

The Infant Hearing Act requires that rules and regulations be adopted and promulgated if the annual percentage rate of newborns who receive a hearing screening during birth admission is less than 95% by December 1, 2003, or at any time thereafter. Hearing screening results reported for occurrent births in 2010 show that 25,925 newborns, or 99.5% of hospital births, were screened during birth admission or prior to discharge from the hospital. The number of newborns screened during birth admission has increased dramatically since reporting began in 2000, when only slightly more than one-third of newborns received a hearing screening during birth admission (see Table 3). This increase in the numbers of newborns receiving a hearing screening corresponds to the

increase in the number of hospitals adopting newborn hearing screening as the standard of care for newborns and the support of sub-grants through the Nebraska Health Care Cash Fund to purchase screening equipment in 2002 and 2003.

Newborns Receiving an Inpatient Hearing Screening (2001-2010)

	2001	2002	2003	2004	2005	2006	2007	2008	2009	2010
Number Receiving a Hearing Screening during Birth Admission	15,272	22,615	25,275	25,966	26,179	26,615	26,737	26,772	26,806	25,923
Percent Receiving a Hearing Screening during Birth Admission	61%	89%	97%	98%	99%	99%	99%	99%	99%	99%

Table 3

Newborns Discharged Without a Hearing Screening

During 2010, the ERS-II reports available for each birthing facility indicated that there were 241 newborns who did not receive a hearing screening during birth admission because the newborn expired prior to screening (100) or were discharged from hospital without a screening (141).

Birth Admission "Refer" Rates

The ERS-II reports available for each birthing facility indicated that 1,129 newborns did not pass the hearing screening during birth admission or prior to discharge from hospital for those babies who were transferred to another hospital. Of the hearing screenings conducted during birth admission, the "refer" rate for all birthing facilities was 4.4% during 2010 (see Table 4). The 2010 refer rate is higher than the refer rates over the last seven (7) years.

Birth Admission "Refer" Rates (2003-2010)

	2003	2004	2005	2006	2007	2008	2009	2010
"Refer" rate for birthing facilities	3.6%	3.5%	3.4%	3.8%	3.7%	4.0%	4.3%	4.4%

Table 4

There are two measurement techniques used to conduct newborn hearing screening: Otoacoustic Emissions (OAE) and Auditory Brainstem Response (ABR). Half of the birthing hospitals in Nebraska are using OAE-only, almost one third are using ABR-only, and the remaining birthing hospitals are using a 2-step method (OAE, followed by ABR if the initial

screening is a "refer"). The "refer" rates differ for the three techniques with the OAE-only having the highest "refer" rate (see Table 5).

"Refer" Rates for Hearing Screening Techniques (2010)

	OAE-only	ABR-only	2-Step
Number of Birthing Facilities	29	24	6
"Refer" Rate	11.1%	2.4%	4.6%

Table 5

Monitoring, Intervention, and Follow-up Care

Another ERS-II report available for each birthing facility is the number of newborns recommended for monitoring, intervention, and follow-up care. In 2010, 649 (57.5% of the babies who did not pass) were recommended for monitoring, intervention and follow-up care by the birthing facilities. Regardless of whether the hospital indicated a recommendation had been made to the parent(s), the NE-EHDI Program's tracking and follow-up processes were followed for each baby who did not pass the hearing screening during birth admission.

The NE-EHDI Program also tracked 1,597 newborns who were transferred to neonatal intensive care units or to hospitals offering a higher level of care in Nebraska and also to surrounding states prior to receiving a hearing screening.

Out-of-Hospital Births

Although parent education material was provided by the NE-EHDI Program to the parents of all reported out-of-hospital births during 2010 who were not immediately admitted to a hospital, only 28% of out-of-hospital births were screened (see Table 6). The remainder were not screened or the results were not submitted to NE-EHDI Program.

Out-of-Hospital Births (2002 – 2010)

	2002	2003	2004	2005	2006	2007	2008	2009	2010
Out-of-hospital births	99	70	60	55	68	77	82	95	94
Number screened	16	12	13	15	30	34	39	34	26
Percentage screened	16	17%	22%	27%	44%	44%	48%	36%	28%

Table 6

Confirmatory Testing/Audiologic Data Reported for 2010

The Advisory Committee for the NE-EHDI Program identified the initial level of the follow-up hearing test for many newborns as an outpatient screening of the newborn's hearing. For those newborns and infants who pass this initial level of follow-up, no further audiologic evaluation would be needed, unless there are risk factors present that would warrant periodic monitoring.

Since the majority of newborns will pass this second screening, considerable cost savings can result by using either the OAE and/or ABR screening technique rather than proceeding directly to a complete audiologic diagnostic evaluation. The Advisory Committee's "Audiological Diagnostic Protocol" recommends that the outpatient screening facility should be prepared to provide comprehensive audiological diagnostic procedures if the outpatient screening results indicate a "refer" status. However, many communities that do not have pediatric audiology services readily available have opted to have the second screening occur at the birthing facility on an outpatient basis.

Type and Degree of Hearing Loss

Neb. Rev. Stat. §71-4739 requires confirmatory testing facilities to report the following:

- Newborns and infants who are shown to have a hearing loss based upon the follow up hearing test.

Analysis of the individually-identifiable confirmatory testing reports submitted to the NE-EHDI Program by July, 2011 indicates 51 infants with a permanent hearing loss in one or both ears. Of that number, 49 of the infants have a hearing loss meeting the criteria for a Permanent Congenital Hearing Loss (PCHL) and two who passed the newborn hearing screening were later identified with a permanent hearing loss (PHL), either acquired or later-onset. Forty (40) of the infants were identified with a bilateral hearing loss, 49% in the mild to moderate range and 51% in the severe to profound range. Eleven (11) infants were identified with a unilateral hearing loss. In the unilateral group, three (3) have atresia and three (3) have auditory neuropathy.

Type and Degree of Permanent Congenital Hearing Loss, 2010 (n = 51)

Degree ► / Type ▼	Bilateral Mild–Moderate	Bilateral Severe–Profound	Unilateral Mild–Moderate	Unilateral Severe-Profound
Sensorineural	13	16	2	3
Conductive	1	-	0	-
Mixed	4	4	0	0
Undetermined	1	0	0	0
Auditory Neuropathy (Disorder)		1	3	
Atresia		0	3	

Table 7

The estimates of the incidence of PCHL in newborns range between one to three per thousand births nationally. Based on the birth rate in Nebraska during 2010 (26,247 including both birthing facility and out-of-hospital births), an estimated 26 to 79 newborns would be identified with PCHL. The incidence of PCHL in Nebraska for babies screened in 2010 reported in individual reports is 1.9 per thousand births.

Tracking and Follow-up Results for 2010

The NE-EHDI Program tracked 1,270 newborns reported as not passing a newborn hearing screening during birth admission. Of those not passing, 1,129 newborns had a birth admission hearing screening "refer" status and 141 newborns were discharged from hospital prior to receiving a hearing screening. These were the newborns tracked through follow up outpatient screenings, diagnostic evaluations and early intervention services.

Follow-up Services and Outcomes

Based on individual reports submitted to the NE-EHDI Program for hospital births, follow-up screening and/or diagnostic evaluations were completed for 1,090 infants with 1,041 having normal hearing and 49 being diagnosed with a PCHL. The evaluation process is still in progress for 38 infants, the parents of seven infants refused to complete the recommended follow up, two families moved with no further contact possible, and eight infants expired before completion of the follow-up services. There were 125 newborns needing follow-up for whom follow up services were not initiated, were initiated but not completed, or were not reported to NE-EHDI Program. These are designated as "Lost to System."

Diagram 1 tracks the services and outcomes of the 1,270 newborns born in 2010 needing follow-up services through the EHDI system and indicates the results for those infants.

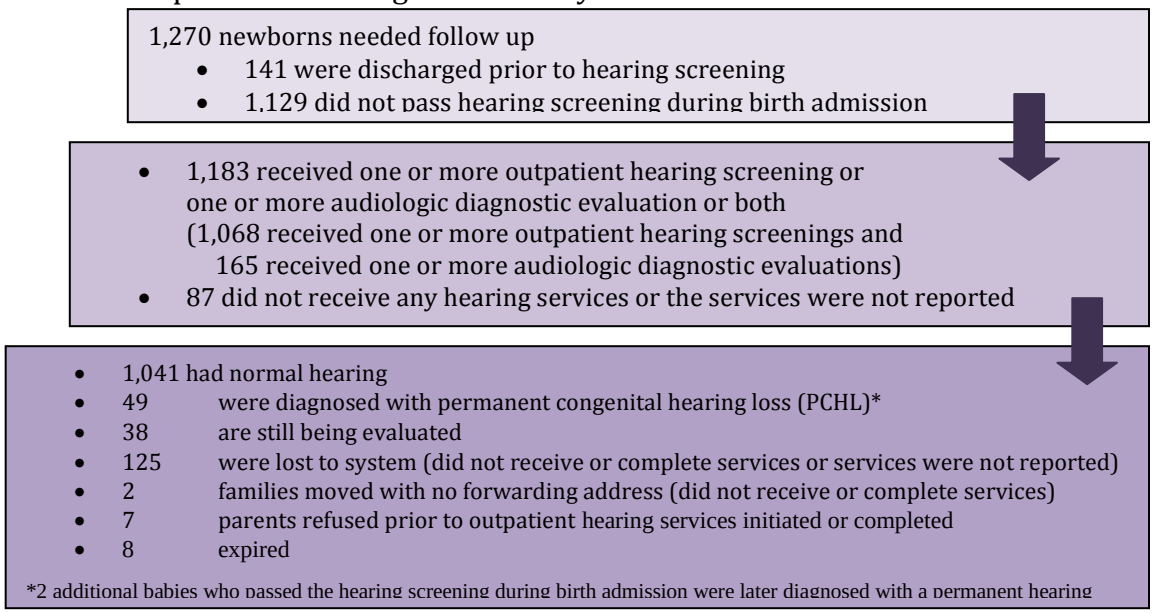


Diagram 1

Timeliness of Follow-up Screening/Testing

To meet the state and national guidelines of "1-3-6" (hearing screening completed by 1 month, audiologic diagnostic evaluation completed by 3 months, early intervention initiated by 6 months), the timeliness of initiation and completion of follow up activities is an important aspect of the quality of services. For the newborns who received follow-up services, 67.1% received an outpatient screening or diagnostic evaluation prior to 1 month of age. Over 94% received follow-up services prior to 90 days of age, the recommended benchmark.

The peak of follow-up activity occurred at approximately 2 weeks of age (see Chart 1). The average age of follow-up service initiation was 32.0 days.

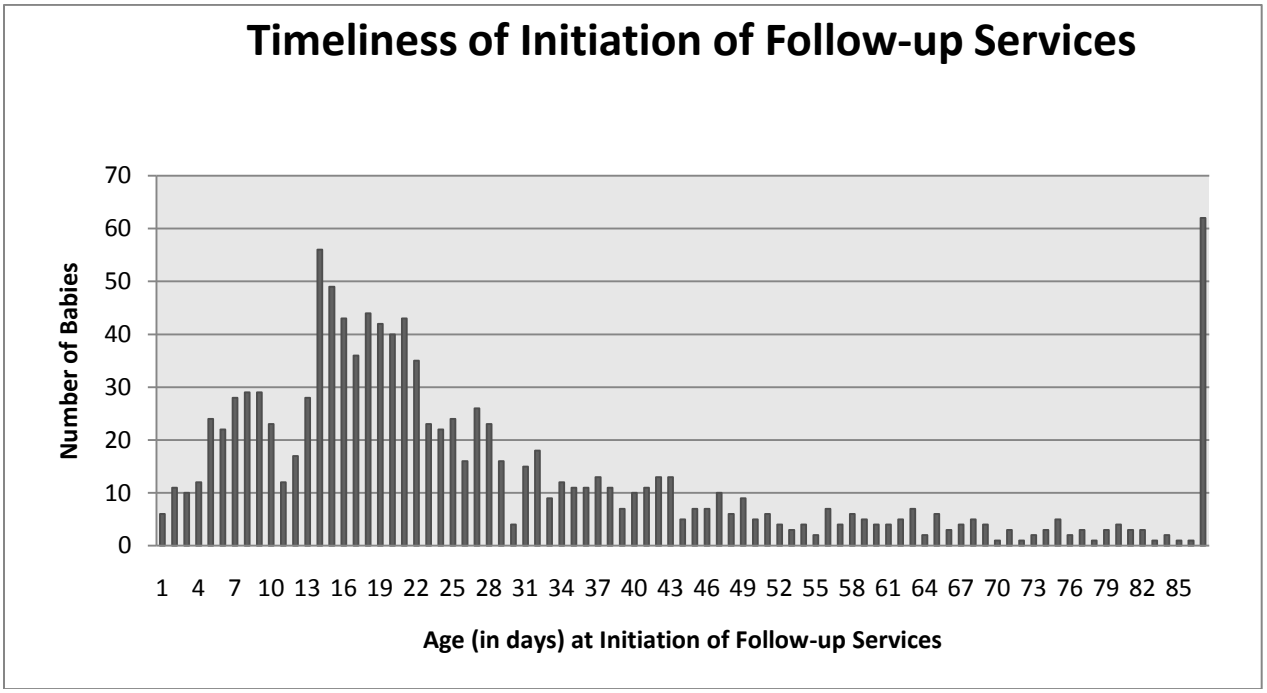


Chart 1

Individual reports were received by the NE-EHDI Program for 51 infants diagnosed with a permanent hearing loss. Two of these were newborns who had passed the newborn hearing screening during birth admission but were diagnosed with a later-onset or progressive hearing loss. The average age at confirmatory diagnosis was 125.2 days with 49% identified prior to three months of age.

Incomplete Results

Neb. Rev. Stat. §71-4742 states: "…it is the goal of this state to achieve a one-hundred-percent screening rate." While Nebraska continues to make very good progress in developing a comprehensive early hearing detection and intervention system, there are also infants for whom the status of their hearing is not known. Overall in 2010, there were 350 babies (including non-hospital births) whose hearing status has not been objectively established:

- 125 infants with no outpatient follow-up initiated, completed or reported after not passing or receiving the inpatient newborn hearing screening.
- 34 infants were identified with hearing problems associated with middle ear dysfunction but additional follow-up evaluations have not yet been completed.
- 62 of the out-of-hospital births were not screened or the results were not submitted to NE-EHDI Program.
- Two families moved before hearing services were initiated or completed.
- Nine parents refused the hearing screening.
- Nine infants with inconclusive results or multiple health problems.

- 109 newborns expired prior to receiving an inpatient hearing screening or completing outpatient follow-up services.

Based on the analysis of the hearing screening and follow-up records, the hearing status (normal hearing or permanent hearing loss) of 98.7% of the 26,247 newborns born in birthing facilities and out-of-hospital has been established.

Early Intervention

The purpose of the Infant Hearing Act (Neb. Rev. Stat. §71-4735) is to "obtain needed multidisciplinary evaluation, treatment, and intervention services at the earliest opportunity and to prevent or mitigate the developmental delays and academic failures associated with late detection of hearing loss." Records for the Early Development Network (EDN), Nebraska's Part C Early Intervention Program, indicate that 46 (90%) of the 51 infants born in 2010 and diagnosed with a PHL (49 PCHL) were referred to EDN. Of the 46 infants referred to EDN, the parent(s) of six infants refused to complete the verification process, three cases did not qualify for EDN services and one case is pending. Of the 36 qualifying for EDN services, 31 (86%) were verified prior to six months of age and five (14%) infants were verified after six months of age.

ACTIVITIES – 2010

Funding

The NE-EHDI Program received funding from the Health Resources Services Administration/ Maternal and Child Health Bureau (HRSA/MCHB) and the Centers for Disease Control and Prevention (CDC). The HRSA/MCHB grant funded the basic operations of the NE-EHDI Program. The CDC cooperative agreement funding supported the development and implementation of the integrated electronic data reporting and tracking system.

Advisory Committee

The NE-EHDI Program was developed based on the requirements identified in the Infant Hearing Act of 2000 and the recommendations by the NE-EHDI Advisory Committee. Specific tasks to be accomplished by the Advisory Committee are 1) to continue to increase the representation of stakeholders, 2) to review and, as necessary, revise the existing protocols to incorporate the electronic data system, 3) to develop new reporting, tracking, and follow-up protocols to effectively link the NE-EHDI Program and the early intervention systems, 4) to increase the program's responsiveness to the expanding cultural and linguistic communities in the state, 5) to support the development of an effective professional development system, and 6) to guide the long-term planning and evaluation of the NE-EHDI system in the state. The Advisory Committee of the NE-EHDI Program consists of 22 members representing medical, audiology, parents, public health, family support, and education stakeholders. The Advisory Committee met three times during 2010. There are three official sub-committees of the NE-EHDI Advisory Committee: Audiology, Evaluation, and Family Support.

Projects

Hearing Screening Equipment for Birthing Facilities

Opportunities to contract for partial funding of new hearing screening equipment were offered to birthing facilities to reduce the number of babies who referred during birth admission hearing screening due to use of aging or inappropriate hearing screening equipment. The funding was made available to small birthing facilities with less than 500 births, large birthing facilities with more than 500 births that were using OAE hearing screening technology, and hospitals with NICUs that did not have dedicated ABR hearing screening equipment.

Electronic Data System

The ERS-II data system was revised to improve functionality and to incorporate additional reports for the birthing facility users. The administrative tracking and follow-up system was also further developed.

Family-to-Family Support

The Family Support Work Group of the NE-EHDI Advisory Committee provided input regarding parent education materials and planning for family support activities. Partnership with the Nebraska chapter of Hands and Voices continued, including exploration of establishing a mentoring program to provide parent-to-parent support when a young child is identified with a permanent hearing loss.

Loss and Found DVD

With funding from the HRSA/MCHB supplemental grant, the NE-EHDI Program joined seven other states in contracting with the national chapter of Hands & Voices to develop an educational DVD for birthing facilities to show to parents of babies who did not pass the newborn hearing screening to reduce the number of babies who are lost to system. The DVD, available in English and Spanish, is based on experiences of families with following up on failed newborn hearing screenings. It includes Nebraska-specific information.

Nebraska Children's Hearing Aid Loaner Bank (NCHALB)

The NCHALB began providing loaner hearing aids to young children in January, 2008. The NCHALB is a partnership between the University of Nebraska - Lincoln Barkley Center, Nebraska Association for the Education of Young Children and the NE-EHDI Program. The NE-EHDI program provides funds to administer the NCHALB and to purchase loaner hearing aids. Requests for additional funding were sent to potential funders. Thirty children were fitted with 52 hearing aids in 2010 and 19 children returned 32 aids.

Summary

- All the current birthing hospitals in Nebraska were conducting newborn hearing screening in 2010. All but one had conducted the hearing screenings during the

birth admission.

- The benchmark of 95% of newborns having a hearing screening during birth admission by December 1, 2003 established by Neb. Rev. Stat. §71-4742 continues to be met. In 2010, birthing hospitals reported screening the hearing of 99% of newborns during birth admission or prior to discharge home for those babies who were transferred to another hospital.
- The overall "refer" rate during 2010 for initial hearing screening during birth admission was 4.4%.
- In 2010, follow-up hearing screenings or audiologic evaluations were initiated within one month of birth for 67.1% of those newborns for which follow-up activities were provided.
- The average age at the time of the initiation of follow-up hearing screening or diagnostic evaluation was 32.0 days.
- For the 51 infants identified with a permanent hearing loss, including congenital, later-onset and progressive hearing loss, the average age at confirmation of hearing loss was 125.2 days.
- There are 350 babies born in 2010 whose hearing was not objectively established and there are 109 who expired before receiving or completing a hearing screening.
- The incidence of Permanent Congenital Hearing Loss identified and reported to NE-EHDI Program (1.9 per thousand screened in 2010) is within the anticipated range of one to three per thousand.
- Over 70% of the infants with a permanent congenital hearing loss were verified for early intervention and special education services.

The staff of the **Nebraska Newborn Screening (Blood-spot) Program** are available to help with your questions at the numbers listed below. General areas of responsibilities are listed:

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This report was prepared and published by the Nebraska Department of Health and Human Services, Newborn Screening Program, P.O. Box 95026, Lincoln, NE 68509-5026. Funding for this report was made possible through the Maternal and Child Health, Title V Block Grant,

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